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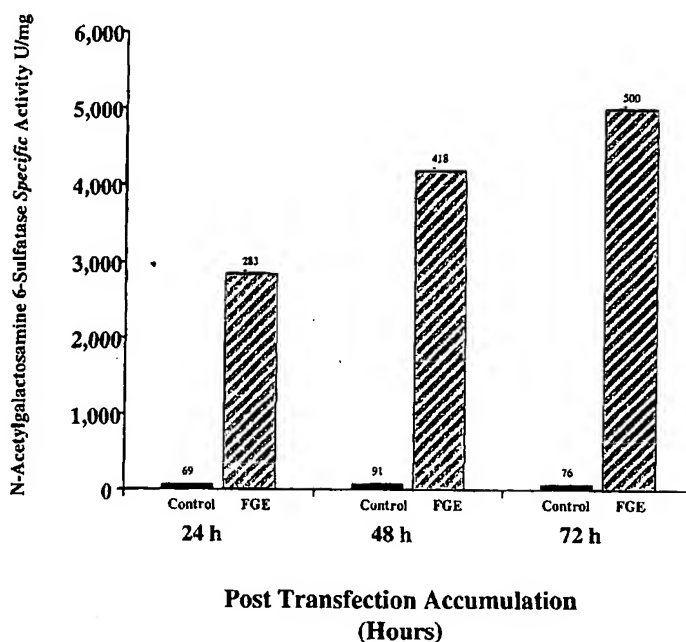
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[Continued on next page]

(54) Title: **DIAGNOSIS AND TREATMENT OF MULTIPLE SULFATASE DEFICIENCY AND OTHER USING A FORMYL-  
GLYCINE GENERATING ENZYME (FGE)**



(57) Abstract: This invention relates to methods and compositions for the diagnosis and treatment of Multiple Sulfatase Deficiency (MSD) as well as other sulfatase deficiencies. More specifically, the invention relates to isolated molecules that modulate post-translational modifications on sulfatases. Such modifications are essential for proper sulfatase function.



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## INTERNATIONAL SEARCH REPORT

International Application No

PCT/US2004/003632

## A. CLASSIFICATION OF SUBJECT MATTER

IPC 7. C12N9/02 C12N15/52 C12Q1/68 A61K38/36 A61K38/44  
A61K31/7088 G01N33/68

According to International Patent Classification (IPC) or to both national classification and IPC

## B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12N C12Q G01N A61K

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

EPO-Internal, BIOSIS, Sequence Search, EMBASE, WPI Data, PAJ

## C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	WO 01/60991 A (WALSH RODERICK T ; INCYTE GENOMICS INC (US); PATTERSON CHANDRA (US); T) 23 August 2001 (2001-08-23)  SEQ ID NOs.: 3 and 14 pages 1-14 page 60 page 82 - page 84; claims 1-28	1-31, 40-42, 56, 58-61, 71-76
Y	SCHIRMER ANDREAS ET AL: "Computational analysis of bacterial sulfatases and their modifying enzymes" CHEMISTRY AND BIOLOGY (LONDON), vol. 5, no. 8, August 1998 (1998-08), pages R181-R186, XP002295627 ISSN: 1074-5521 the whole document	1-85

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## INTERNATIONAL SEARCH REPORT

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PCT/US2004/003632

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
Y	FEY JENS ET AL: "Characterization of posttranslational formylglycine formation by luminal components of the endoplasmic reticulum" JOURNAL OF BIOLOGICAL CHEMISTRY, vol. 276, no. 50, 14 December 2001 (2001-12-14), pages 47021-47028, XP002295628 ISSN: 0021-9258 cited in the application the whole document	1-85
X	----- DATABASE EMBL 18 September 2002 (2002-09-18), XP002296298 Database accession no. BD551115 the whole document	19,20, 58-61
X	----- DATABASE EMBL 26 March 2002 (2002-03-26), XP002296299 Database accession no. ABB62912 the whole document	19,20, 58-61
X	----- DATABASE EMBL 1 October 2001 (2001-10-01), XP002296300 Database accession no. Q98BQ8 the whole document	19,20, 58-61
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X	----- DATABASE EMBL 23 May 2001 (2001-05-23), XP002296302 Database accession no. AAB88402 the whole document	19,20, 58-61
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## INTERNATIONAL SEARCH REPORT

International Application No

PCT/US2004/003632

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	DATABASE EMBL 1 May 1997 (1997-05-01), XP002296306 Database accession no. P95060 the whole document -----	19,20, 58-61
X	DATABASE EMBL 1 December 2001 (2001-12-01), XP002296307 Database accession no. Q92WL9 the whole document -----	19,20, 58-61
X	DATABASE EMBL 1 December 2001 (2001-12-01), XP002296308 Database accession no. Q93PA2 the whole document -----	19,20, 58-61
X	DATABASE EMBL 1 June 2001 (2001-06-01), XP002296309 Database accession no. Q9A921 the whole document -----	19,20, 58-61
A	SCHMIDT BERNHARD ET AL: "A Novel Amino Acid Modification in Sulfatases That Is Defective in Multiple Sulfatase Deficiency" CELL, vol. 82, no. 2, 1995, pages 271-278, XP002295629 ISSN: 0092-8674 cited in the application the whole document -----	
P,X	DATABASE EMBL 1 October 2003 (2003-10-01), XP002296310 Database accession no. Q7V5N5 the whole document -----	19,20, 58-61
P,X	DATABASE EMBL 1 March 2003 (2003-03-01), XP002296311 Database accession no. Q8FTJ8 the whole document -----	19,20, 58-61
P,X	DATABASE EMBL 1 June 2003 (2003-06-01), XP002296312 Database accession no. Q88HK3 the whole document -----	19,20, 58-61
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## INTERNATIONAL SEARCH REPORT

International Application No

PCT/US2004/003632

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
P,X	<p>DIERKS THOMAS ET AL: "Multiple sulfatase deficiency is caused by mutations in the gene encoding the human Calpha-formylglycine generating enzyme." CELL, vol. 113, no. 4, 16 May 2003 (2003-05-16), pages 435-444, XP002295630 ISSN: 0092-8674 the whole document</p> <p>-----</p>	1-85

## INTERNATIONAL SEARCH REPORT

International application No.  
PCT/US2004/003632

### Box II Observations where certain claims were found unsearchable (Continuation of Item 2 of first sheet)

This International Search Report has not been established in respect of certain claims under Article 17(2)(a) for the following reasons:

1. ☒ Claims Nos.:  
because they relate to subject matter not required to be searched by this Authority, namely:  
see FURTHER INFORMATION sheet PCT/ISA/210
2. ☐ Claims Nos.:  
because they relate to parts of the International Application that do not comply with the prescribed requirements to such an extent that no meaningful International Search can be carried out, specifically:
3. ☐ Claims Nos.:  
because they are dependent claims and are not drafted in accordance with the second and third sentences of Rule 6.4(a).

### Box III Observations where unity of invention is lacking (Continuation of Item 3 of first sheet)

This International Searching Authority found multiple inventions in this international application, as follows:

1. ☐ As all required additional search fees were timely paid by the applicant, this International Search Report covers all searchable claims.
2. ☐ As all searchable claims could be searched without effort justifying an additional fee, this Authority did not invite payment of any additional fee.
3. ☐ As only some of the required additional search fees were timely paid by the applicant, this International Search Report covers only those claims for which fees were paid, specifically claims Nos.:
4. ☐ No required additional search fees were timely paid by the applicant. Consequently, this International Search Report is restricted to the invention first mentioned in the claims; it is covered by claims Nos.:

#### Remark on Protest

- ☐ The additional search fees were accompanied by the applicant's protest.
- ☐ No protest accompanied the payment of additional search fees.



**FURTHER INFORMATION CONTINUED FROM PCT/ISA/ 210**

**Continuation of Box II.1**

Although claims 21-31, 33-39 and 57 are directed to or may comprise a diagnostic method practised on the human/animal body (e.g. the provision of a biological sample is a step to be practised on the human or animal body), the search has been carried out and based on the alleged effects of the compound/composition.

Although claims 44-55, 62-67 and 82-85 are directed to a method of treatment of the human/animal body, the search has been carried out and based on the alleged effects of the compound/composition.

## INTERNATIONAL SEARCH REPORT

International Application No  
PCT/US2004/003632

Patent document cited in search report		Publication date	Patent family member(s)	Publication date
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			CA 2400034 A1	23-08-2001
			EP 1255819 A2	13-11-2002
			JP 2004502404 T	29-01-2004
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